



PNKD gene

paroxysmal nonkinesigenic dyskinesia

Normal Function

Researchers have not determined the role of the *PNKD* gene (frequently called the *MR1* gene) in the human body. This gene is highly active (expressed) in the brain, which suggests that it plays an important role in normal brain function. The protein produced from the *PNKD* gene is similar to another protein that helps break down a chemical called methylglyoxal. Methylglyoxal is found in alcoholic beverages, coffee, tea, and cola. Research has demonstrated that this chemical is toxic to nerve cells (neurons). The *PNKD* protein is expected to perform a function similar to this known protein.

Health Conditions Related to Genetic Changes

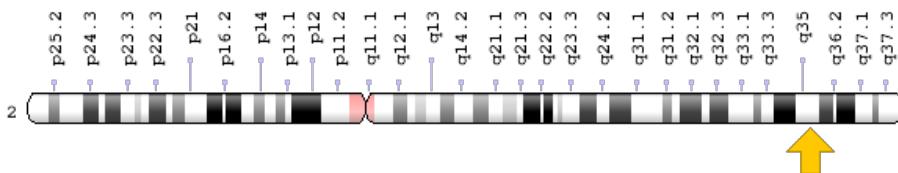
familial paroxysmal nonkinesigenic dyskinesia

At least two mutations in the *PNKD* gene have been shown to cause familial paroxysmal nonkinesigenic dyskinesia. Both mutations replace the protein building block (amino acid) alanine with the amino acid valine in the *PNKD* protein. One of the mutations occurs at position 7 (written as Ala7Val or A7V), and the other mutation is found at position 9 (written as Ala9Val or A9V). Research suggests that these mutations alter the structure of the *PNKD* protein and interfere with its ability to function. It is not known how mutations in the *PNKD* gene lead to the signs and symptoms of familial paroxysmal nonkinesigenic dyskinesia.

Chromosomal Location

Cytogenetic Location: 2q35, which is the long (q) arm of chromosome 2 at position 35

Molecular Location: base pairs 218,270,392 to 218,346,793 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- brain protein 17
- BRP17
- DKFZp564N1362
- DYT8
- FKSG19
- FPD1
- KIAA1184
- KIPP1184
- MGC31943
- MR-1
- MR1
- myofibrillogenesis regulator 1
- PDC
- PKND1
- PNKD_HUMAN

Additional Information & Resources

GeneReviews

- Familial Paroxysmal Nonkinesigenic Dyskinesia
<https://www.ncbi.nlm.nih.gov/books/NBK1221>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PNKD%5BTIAB%5D%29+OR+%28paroxysmal+nonkinesigenic+dyskinesia%5BTIAB%5D%29%29+OR+%28MR-1%5BTIAB%5D%29+OR+%28MR1%5BTIAB%5D%29+OR+%28myofibrillogenesis+regulator+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28%28histocompatibility%5BTIAB%5D%29+OR+%28Doxepin%5BTIAB%5D%29+OR+%28MHC%5BTIAB%5D%29+OR+%28HLA%5BTIAB%5D%29+OR+%28immunotoxin%5BTIAB%5D%29+OR+%28mecA%5BTIAB%5D%29+OR+%28autoimmune%5BTIAB%5D%29+OR+%28HeLa%29+OR+%28Immunomodulation%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- MYOFIBRILLOGENESIS REGULATOR 1
<http://omim.org/entry/609023>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PNKD.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PNKD%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9153
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/25953>
- UniProt
<http://www.uniprot.org/uniprot/Q8N490>

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